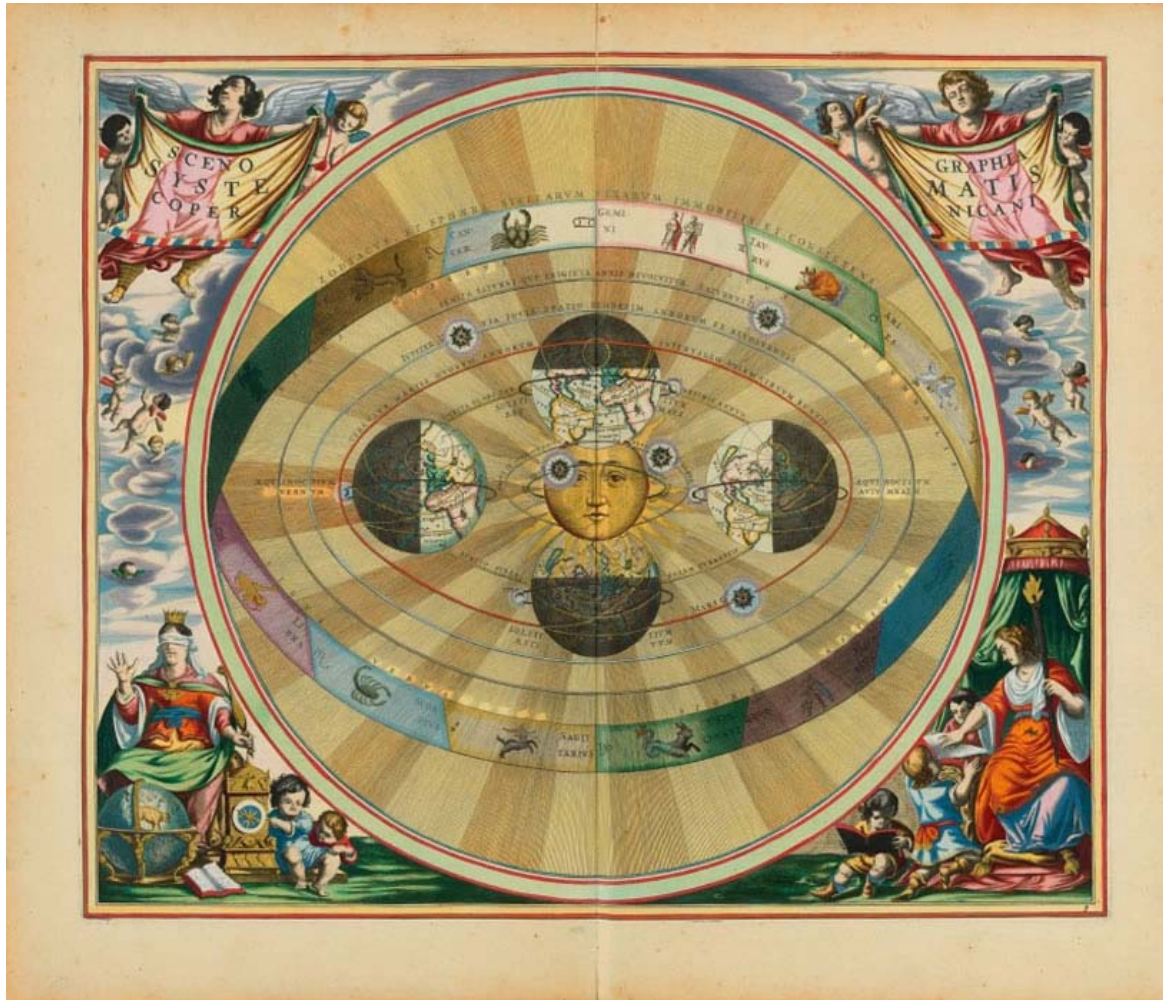
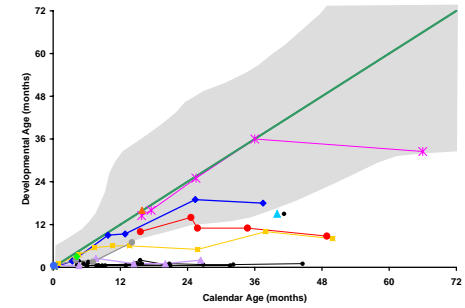
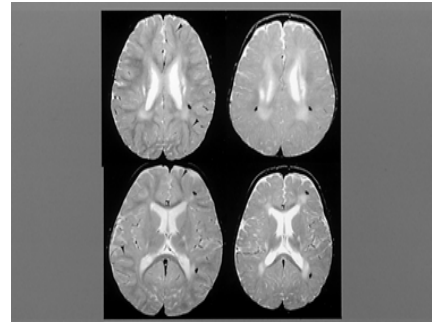
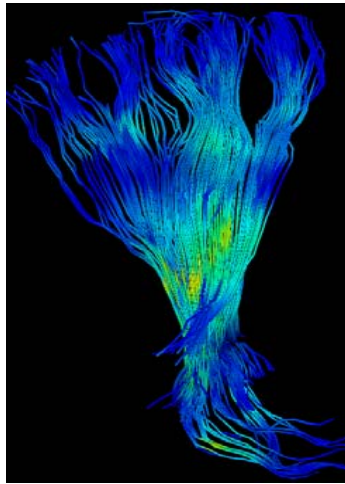


Linear mentoring evolves into networks of scientists



Janet Rubin
Margaret
Gourlay
Maria Escolar



UCBT

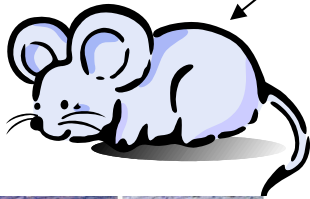


FIGURE 4. Pathology of the peripheral nerve. (A) SAP-4^{-/-} mouse and (B) Chimeric SAP-4^{+/+} mouse, both at PND 190, LFB-PAS stains. (C) Chimeric SAP-4^{+/+} mouse at PND 310, LFB-PAS stain. (D) Chimeric SAP-4^{+/+} mouse at PND 310, GFP immunocytochemistry. (E) SAP-4^{-/-} mouse and (F) Chimeric SAP-4^{+/+} mouse, both at PND 190 on one-quarter-thick epon sections. (G) Chimeric SAP-4^{+/+} mouse at PND 310. Single arrows and arrowheads indicate nerve fibers with thin myelin sheaths and onion bulb formation respectively. Double arrows in (F) indicate normal appearing myelinated fibers. An electron micrograph of the sciatic nerve (H) of a chimeric SAP-4^{+/+} mouse at PND 310 shows many thin cellular processes forming onion bulbs. Scale bars = (A-G) 100 μ m; (H) 5 μ m.

Mentor # 1

- Dr. Joanne Kurtzberg, Pediatric Bone Marrow Transplantation at Duke University Medical Center



Contracts a small percentage of my clinical time

Encourages clinical research

Meets regularly with me

I observe her grow as a leader in her field

Mentor # 2

- Dr. Mel Levine, Director for the Center of Development and Learning at UNC



Provides space and initial resources

Recognizes the importance of my work

Challenges me to become independent

The NFRD

- Where is the funding?
- Multidisciplinary team
- Clinical service
- No time for research

Krabbe Disease



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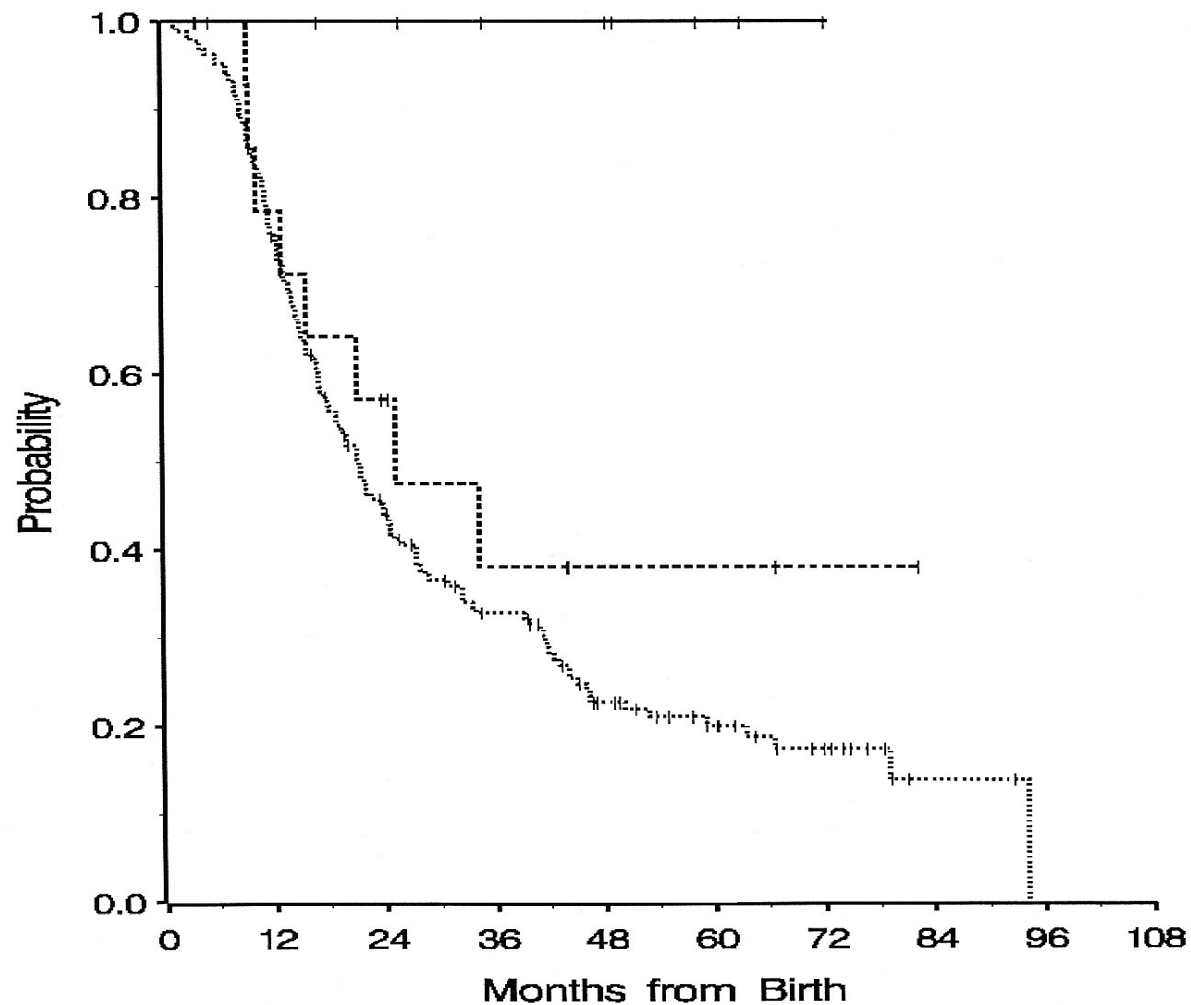
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Transplantation of Umbilical-Cord Blood in Babies with Infantile Krabbe's Disease

Maria L. Escolar, M.D., Michele D. Poe, Ph.D., James M. Provenzale, M.D., Karen C. Richards, M.D., June Allison, R.N., Susan Wood, P.N.P., David A. Wenger, Ph.D., Daniel Pietryga, M.D., Donna Wall, M.D., Martin Champagne, M.D., Richard Morse, M.D., William Krivit, M.D., Ph.D., and Joanne Kurtzberg, M.D.

EVENT – FREE SURVIVAL

- Newborns Transplanted with UCBT for Krabbe Disease (N=11)
- - - Symptomatic Patients Treated with UCBT (N=14)
- Untreated Control Group (Natural History N=190)



August 2006

**Newborn Screening for Krabbe
Disease is implemented in New
York State**

The goal of the NFRD program is to investigate the course of rare diseases over time and understand how they impact the brain and the nervous system.

What is NFRD?

N **Neurodevelopment** describes how the brain and nervous system change over time.



Why is this important?

Improving our understanding of neurodevelopment allows us to devise novel treatments and interventions to enhance the patients' quality of life.

F **Function** describes how children interact and adapt to their environment.

Why is this important?

These diseases may affect function by interfering with the rate at which children acquire the skills needed for independence in their daily lives. Addressing these developmental differences allows us to design specific interventions to improve their ability to function independently.



R **Rare** disorders are not commonly identified in the population. Because of the low incidence of these diseases it has been difficult to gather enough information to understand how the disease affects all aspects of the child's life.

Why is this important?

By gathering information about these disorders we can understand the specific impact of the disease and the most effective interventions.



D **Disorders** refers to the specific abnormalities associated with each disease. By evaluating children with a various rare diseases gives us insight into the unique aspects of each condition.

Why is this important?

These diseases affect multiple systems, therefore interventions must address all aspects of the child's disorder.



History of NFRD Program

The NFRD was started in 2000 because of the need to help children and their families understand the overall impact of genetic, neurodegenerative diseases. While the program initially focused on lysosomal storage disorders, it has now expanded to include over 20 rare inherited disorders.

Mentor # 3

Dr. Joseph Piven, Director of the Neurodevelopmental Research Center (NDRC) at UNC



Common research interests

Has experience with NIH grants

Identifies key resources within UNC

Serves as a role model

Mentor # 4

- Dr. Eugene Orringer



Provides funding to protect research time

Identifies mentors at different levels

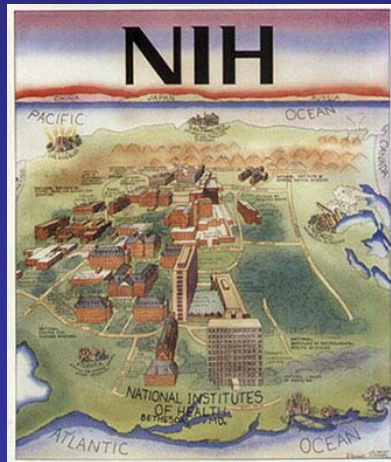
Supports all aspects of my work (clinical, research, academic)

Available when crisis strikes!

My first NIH grant – R01

DTI as a tool to identify babies with Krabbe Disease in need of urgent treatment

In a prospective study of 100 babies with low GALC enzyme, determine if DTI can identify which newborns will develop infantile Krabbe Disease



Mentor # 5

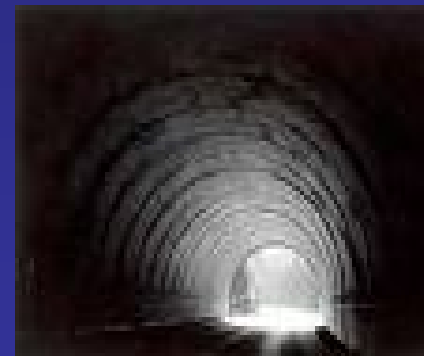
- Dr. Janet Rubin



“Don't despair. IT'S ALL GOOD.

No one can take away
SIGNIFICANCE, NOVELTY and
IMPACT away from you.

You go girl”.



Mentor # 6

- Dr. Margaret Gourley



"Today I had to see patients until late, and work on some issues we had with the budget. I still need to send drafts of letters, look at the abstract, environment and data dissemination. Grrrrrr" Maria



I can put in a few hours of work after 8:30 tonight. Overall, you are in great shape. I will be on call on Friday night and in clinic on Saturday. Although I might have to take a nap first, I can help you late Saturday PM. On Sunday, you can do the page count on the full file so we know how close it is to 25 pp. Then you can trim and refine." Margaret

R01 Funded!